



# SPINAL MUSCULAR ATROPHY

## Early Diagnosis and Access to Treatment Saves Lives!

Diagnostic Delays in SMA are common. However, early provision of treatment is critical to modifying the rapid and irreversible loss of motor neurons and exponentially increasing chances of survival and functional gains to patients.

Disease-modifying therapies are available for SMA and early treatment has a substantial beneficial impact on clinical outcomes.



## WHAT IS SMA?

- Autosomal recessive neuromuscular disease caused by deletion/mutation in SMN 1 gene
- Hypotonia and weakness in the trunk and limbs are the predominant symptoms
- Symptoms present with acute onset and rapid deterioration followed by a prolonged plateau
- One of the most common monogenic disorders in humans

## CLINICAL PRESENTATION

SMA has historically been categorized into subtypes based on age that symptoms begin and highest motor milestone achieved. This classification continues to apply to adults, teens, and some children in SMA. The most common three types in children present as follows:

### SYMPTOM ONSET < 6 MONTHS

#### *Historically Type 1*

- Hypotonia
- Typical supine position is bent elbows and frog legs
- Difficulty lifting extremities against gravity
- Rapid paradoxical breathing or belly breathing
- Bell-shaped chest
- Poor head control/head lag
- Bulbar weakness/dysphagia
- Never sits

### SYMPTOM ONSET 6 TO 18 MONTHS

#### *Historically Type 2*

- Hypotonia
- Difficulty lifting extremities against gravity
- Possible bell-shaped chest
- Possible paradoxical breathing or belly breathing
- Tongue fasciculations
- Absent deep tendon reflexes
- Delayed sitting and never walks

### SYMPTOM ONSET 1.5 TO 10 YEARS

#### *Historically Type 3*

- Hypotonia
- Muscle tremor
- Fatigue with exertion and frequent falls
- Gower's sign, Trendelenburg gait, hyperlordosis, and overpronation observed
- Delayed walking

*Genetic testing is required to confirm SMA.*

## RESOURCES

- SMA Diagnostic Toolkit: [www.curesma.org/SMARTmoves-toolkit](http://www.curesma.org/SMARTmoves-toolkit)
- SMA Treatment Overview: [www.curesma.org/spinal-muscular-atrophy-treatment/](http://www.curesma.org/spinal-muscular-atrophy-treatment/)



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